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(54) Title: METHOD AND COMPOSITIONS FOR DETECTION AND ENUMERATION OF GENETIC VARIATIONS

(57) Abstract: Many areas of biomedical research depend on the analysis of uncommon variations in individual genes or transcripts. Here we describe a method that can quantify such variation at a scale and ease heretofore unattainable. Each DNA molecule in a collection of such molecules is converted into a single particle to which thousands of copies of DNA identical in sequence to the original are bound. This population of beads then corresponds to a one-to-one representation of the starting DNA molecules. Variation within the original population of DNA molecules can then be simply assessed by counting fluorescently-labeled particles via flow cytometry. Millions of individual DNA molecules can be assessed in this fashion with standard laboratory equipment. Moreover, specific variants can be isolated by flow sorting and employed for further experimentation. This approach can be used for the identification and quantification of rare mutations as well as to study variations in gene sequences or transcripts in specific populations or tissues.